Objective Evaluation of Craniofacial Features in Patients with Down's Syndrome

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Summary

Clinical anthropometry is used for evaluation of the features of the craniofacial region. The aim of this study was to determine the anthropometric variables which best discriminate a young population with Down's Syndrome (DS) from healthy subjects and to produce a craniofacial anthropometric profile (CAP) specific for DS. The study was performed on a sample of 56 subjects (33 male and 23 female) with DS, divided into two age groups (7-12 and 13-18 years). The control group comprised 322 healthy subjects (151 male and 171 female) of the same age as the examined groups. Twenty standard craniofacial measurements were measured with instruments according to Martin and in accordance with Farkas protocol. The measured values of all subjects with DS were expressed by mean and standard deviation. CAP was performed by converting individual measurements to standard values. Deviation from mean values was considered significant when z-value deviated by more than +2 or less than -2. The results showed that the variables which discriminated the group of subjects with DS from healthy subjects in the subnormal area in the first age group were - length of the head, length of the auricles and head circumference, and in the second age group - length of the head, length of the auricles, width of the auricles and head circumference. CAP can be considered a useful and objective method in defining specific craniofacial features of DS.

Key words: *Down's Syndrome, craniofacial features, anthropometric variables, craniofacial anthropometric profile.*

Introduction

Down's Syndrome (DS) is the most frequent chromosomatopathy in man, which is the cause of mental retardation and comprises around 10% of all cases of severe mental retardation (1). Today it is well known that DS is a specific, trisomy 21 defined syndrome with multiple congenital anomalies and mental retardation. The incidence of DS births on average amounts to approximately one case in 600-800 births (2, 3). More than 100 different characteristic signs for DS have been reported in the literature, of which the majority is in the craniofacial region (3-6). The most important common signs of DS are: low stature, mental retardation, hypotonia, congenital cardiac diseases, small brachyphalic head, flat skull, epicantal folds, Brushfield's freckles, cloudy lens, wide nasal ridge, wrinkled, low posi-

tioned, plainly formed ears, small upper jaw, open mouth with a protruding, large, fissured tongue, short neck, arms and fingers, clinodactyly of the fifth digit, dermatoglyphs and a wide space between the first and second toes. A characteristic physical finding of the craniofacial complex includes insufficiently developed middle part of the face, with hypoplastic upper jaw and nasal bones (7). According to J.H.L. Down the two main characteristics of the face in DS were a flat face and small, centrally positioned features (eyes, nose and mouth) (8). In the case of DS the size of the craniofacial complex remains smaller throughout the whole lifetime, compared with a normal, healthy population. However, some changes in growth and with increased age (e.g. less protruding forehead and less retruded upper jaw) contribute to the fact that such patients in adulthood occasionally no longer have clear recognisable clinical features such as those in their youth (4, 6, 9). Because people with DS have several tens of different, characteristic signs in the craniofacial area, clinical anthropometry is one of the methods which can very successfully be used for evaluation of the features of the craniofacial region, in order to avoid subjectivity (10-12). Mere inspection of the head and face, without the application of objective methods of evaluation of the craniofacial region, it is possible for mistakes to be made in subjects with DS because of apparent dysmorphism. Clinical anthropometry has particular value with the introduction of methods of craniofacial anthropometric profile (CAP), which enables comparison of individuals or groups suffering from genetic disease with standard for healthy persons of the same age and sex. The object of this study was to determine, in a representative sample of young patients with DS, the specificity of CAP for the syndrome, which can be used in the diagnosis, monitoring of growth and eventual plastic-surgical procedures in the craniofacial region, and to identify anthropometric variables which best discriminate a group of patients with DS from healthy persons.

Subjects and methods

The study was performed on 56 patients (33 male and 23 female) with DS. A diagnosis of trisomy 21 was determined clinically and by karyotypisation.

The age of the subjects ranged from 7 to 18 years. Subjects with DS were classified into two age groups: 7-12 years and 13-18 years. The healthy control group consisted of 322 persons (151 male and 171 female). The control group was divided into identical age groups as the examined groups. Twenty standard craniofacial measurements, which are important for producing a craniofacial anthropometric profile (CAP), were measured for each subject. Craniofacial variables were: width of the head (eu-eu), width of the forehead (ft-ft), wide of the base of the skull (t-t), upper facial width (zy-zy), width of the nose (al-al), lower facial width (go-go), width of the mouth (ch-ch), length of the head (g-op), upper facial depth (n-t), middle facial depth (sn-t), lower facial depth (gnt), length of the auricles (sa-sba), width of the auricles (pra-pra), length of the nose (n-sn), total facial height (n-gn), internal cantal distance (en-en), external cantal distance (ex-ex), circumference of the head (g-op-g), maxillary surface arch (t-sn-t) and mandibular surface arch (t-gn--t). Measurement was carried out with original, standardised instruments for anthropometric measurements according to Martin (sliding callipers, sliding scale etc.) and in accordance with Farkas protocol (13). Assessments of basic parameters of distribution of variables of the craniofacial anthropometric status for all subjects with DS are expressed by percentage and standard deviation. As a craniofacial anthropometric profile is an inevitable, exceptionally practical segment of anthropometry, it was obtained by converting individual measurements into standardised values (z-values) and thus analysis was made of the standardised values of variables of the craniofacial status of subjects with DS in relation to healthy subjects. Deviation from mean values was considered significant when z-value deviated by more than +2 or less than -2.

Results

The results of the anthropometric study of the head and face of male and female subjects with DS, aged from 7 to 18 years, classified into two age groups (7-12 years and 13-18 years), are presented in Table 1. All measurements are given in the same order and in the same way as in the list of variables. The mean values of almost all variables for the female subjects were less than for the male subjects,

which was the same in the control group. Craniofacial anthropometric profiles of the variables for both male and female subjects with DS, according to age groups compared to the mean and standard deviations of subjects in the control group are presented in Figures 1 and 2. All profiles were arranged so that standardised values are shown on a vertical axis, while the craniofacial variables are aligned on a horizontal axis in the same order as the list of variables, thus creating a unique base for all profiles. The graphic presentation in Figure 1 relates to a comparison of the craniofacial anthropometric profile between male and female subjects with DS within the first age group (7-12 years). This age group of subjects with DS shows three variables which are located in the subnormal area, i.e. (less than -2) and are the same for male and female subjects. They are: length of head (g-op), length of the auricles (sa-sba) and head circumference (circumference). All other variables are located in the normal area (-2 to +2). Female subjects have lower values than male subjects in almost all variables. The following four variables are exceptions: lower facial width (go-go), length of head (g-op), width of nose (al-al) and external cantal distance (ex-ex). Comparison of the craniofacial anthropometric profile between male and female subjects with DS in the second age group (13-18) is shown in Figure 2. In comparison to the first age group with DS, this age group showed one more variable which is included in the subnormal area, i.e. less than -2 and relates to both sexes. These variables are: length of head (g-op), length of auricles (sa-sba), width of auricles (pra-pa) and head circumference. Of the aforementioned variables three were already included in the description of the previous age group, and in this age group width of the auricles (pra-pa) was also included. All other variables fall in the normal area (-2 to +2). The female subjects had lower values than the male subjects in almost all variables (exceptions were: head width (eu-eu), head length (g-op), total facial height (n-gn), width of the mouth (ch-ch) and circumference of the head (circumference).

Discussion

Clinical findings corroborate the theory that the specificities found in persons with DS are the result of irregular development and growth in the early embryonic period (14). In the case of trisomy 21 additional genetic material is known which can "confuse" the normal polygenetic model and thus manifest in generalised and localised growth disorders (13). From the large number of measured anthropometric sizes of the craniofacial complex, sufficient data were obtained in this study for determination of differences between the measured values of healthy subjects and subjects with DS. Allanson et al (9) carried out the only study so far in the world on DS, using CAP, in which similar methodology was used as in the present study. Therefore the use of CAP analysis in a large number of subjects of different age groups is very important for scientific verification of morphological occurrences in the craniofacial region in persons with DS. However, as different population groups were involved (American and Croatian populations) both of healthy subjects and of those with DS, it is impossible to completely compare the results of these two studies. The investigation by Farkas also involved the anthropometry of DS for the purpose of plastic-surgical methods on such persons, and thus the results of this study can be compared with his results (9, 13, 14). In their investigations both Allanson & Farkas used standards for North-American Caucasians (15). Comparison of the North-American norms with our population shows that much less importance is paid to the values for head width (eu-eu), head circumference (circumference), external cantal distance (ex-ex) and width of the nose (al-al) (15). This is important to mention because of interpretation and comparison with the results of this study and the American study. Namely, the heads of Americans are narrower and thus when compared with the group of DS these differences are more emphasised in their investigations (due to the fact that in the case of DS brachyphalia is expected). The results of this study in a group of patients with DS most often show subnormal craniofacial values, which is in agreement with other findings (4, 16-19). The great diversity of results in the literature is due to the fact that different methods of research were frequently used. Namely, clinical and radiographic findings are most frequently mentioned and anthropometric findings least frequently mentioned. In this study the highest incidence of subnormal measurements of subjects with DS was for the length of the head (g-op), head circumference (circumference), width of the auri-

cles (pra-pa) and length of the auricles (sa-sba). This agrees with the results of the analyses of DS carried out by Farkas (13, 20). Decreased head length can be considered brachyphalia in this specific population, while reduced circumference indicates microcephalia. Decreased width and length of the auricles are features which are traditionally said to be discriminative for DS. Head circumference (circumference) cannot be considered the only measurement which determines reduced head size. The length of the head is a measurement which has a direct influence on the head circumference and it is not influenced by the width of the forehead or by the width of the head. This study indicates the possibility of applying anthropometric measurement of variables for producing a craniofacial anthropometric profile, which gives a completely objective picture of that which is determined on the head and face of the patient by a clinical examination and inspection. Developmental changes determined in the present study show that trisomy 21 has a clearly defined and different phenotype than the healthy population as early as childhood. These characteristics are emphasised during adolescence and can be expected to change with age. In the case of trisomy 21, the additional chromosome 21 is clearly the carrier of numerous detected and still undetected genes, whose interactions are still unknown. In spite of achievements and investigations on the level of the gene, the method of anthropometric evaluation of the craniofacial region is still inevitable and can be successfully applied in clinical genetics.

Conclusion

On the basis of anthropometric measurements of the craniofacial system it is possible to conclude that persons with DS have a specific and recognisable CAP with specifically expressed deviations from normal, which enable their discrimination from healthy persons. Analysis of standard values (z-values) of the craniofacial status shows that the majority of variables are located most frequently in the normal area of standardised values (from-2 to +2), and some, characteristic variables for DS, in the subnormal area (from -4 to -2), which means that these values are abnormally small in the case of DS, compared to a healthy population. Comparison of younger and older age groups showed a visible increase in the number of variables in the subnormal area, accompanied by increase in the age of the subjects.